MASP1 gene

mannan binding lectin serine peptidase 1

Normal Function

The *MASP1* gene provides instructions for making proteins that are involved in a series of reactions called the lectin complement pathway. This pathway is thought to help direct the movement (migration) of cells during early development before birth to form the organs and systems of the body. It appears to be particularly important in directing the migration of neural crest cells, which give rise to various tissues including many tissues in the face and skull, the glands that produce hormones (endocrine glands), and portions of the nervous system. After birth, the lectin complement pathway is involved in the immune system.

Three different proteins, MASP-1, MASP-3, and MAp44 can be produced from the *MASP1* gene, depending on how the gene's instructions are pieced together.

Health Conditions Related to Genetic Changes

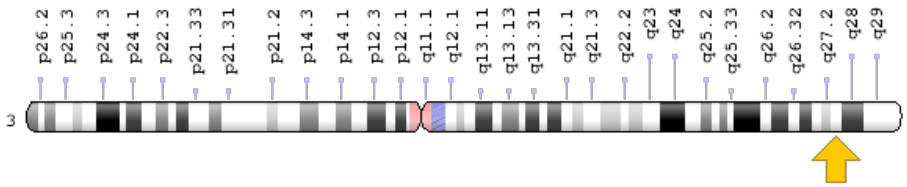
3MC syndrome

At least 10 *MASP1* gene mutations have been identified in people with 3MC syndrome, a disorder characterized by unusual facial features and a variety of problems affecting other tissues and organs of the body. The *MASP1* gene mutations that cause 3MC syndrome affect the MASP-3 protein; some affect the MASP-1 protein in addition to MASP-3. The protein changes result in faulty control of cell migration in embryonic development, leading to the various abnormalities that occur in this disorder. Researchers suggest that the existence of parallel pathways in the immune system that can compensate for problems in the lectin complement pathway account for the absence of immune system problems in 3MC syndrome.

Chromosomal Location

Cytogenetic Location: 3q27.3, which is the long (q) arm of chromosome 3 at position 27.3

Molecular Location: base pairs 187,216,085 to 187,292,220 on chromosome 3 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 3MC1
- complement-activating component of Ra-reactive factor
- complement factor MASP-3
- CRARF
- CRARF1
- mannan-binding lectin serine peptidase 1 (C4/C2 activating component of Ra-reactive factor)
- mannose-binding lectin-associated serine protease 1
- mannose-binding protein-associated serine protease
- MAP1
- MAp44
- MASP
- MASP3
- PRSS5
- Ra-reactive factor serine protease p100
- RaRF
- serine protease 5

Additional Information & Resources

Educational Resources

- Immunobiology (fifth edition, 2001): The mannan-binding lectin pathway is homologous to the classical pathway
  https://www.ncbi.nlm.nih.gov/books/NBK27100/#A170

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28MASP1%5BTIAB%5D%29+OR+%28mannan-binding+lectin+serine+peptidase+1%5BTIAB%5D%29%29+OR+%28%283MC1%5BTIAB%5D%29+OR+%28CRARF%5BTIAB%5D%29+OR+%28MAP1%5BTIAB%5D%29+OR+%28MASP3%5BTIAB%5D%29+OR+%28MASP%5BTIAB%5D%29+OR+%28MAp44%5BTIAB%5D%29+OR+%28Ra-reactive+factor+serine+protease+p100%5BTIAB%5D%29+OR+%28RaRF%5BTIAB%5D%29+OR+%28complement+factor+MASP-3%5BTIAB%5D%29+OR+%28complement-activating+component+of+Ra-reactive+factor%5BTIAB%5D%29+OR+%28mannan-binding+lectin+serine+protease+1+isoform+1+precursor%5BTIAB%5D%29+OR+%28mannan-binding+lectin+serine+protease+1+isoform+2+precursor%5BTIAB%5D%29+OR+%28mannan-binding+lectin+serine+protease+1+isoform+3+precursor%5BTIAB%5D%29+OR+%28mannose-binding+lectin-associated+serine+protease+1%5BTIAB%5D%29+OR+%28mannose-binding+protein-associated+serine+protease%5BTIAB%5D%29+OR+%28serine+protease+5%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D

OMIM

- MANNAN-BINDING LECTIN SERINE PROTEASE 1
  http://omim.org/entry/600521

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_MASP1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=MASP1%5Bgene%5D
- HGNC Gene Family: Proteases, serine
  http://www.genenames.org/cgi-bin/genefamilies/set/738
- HGNC Gene Family: Sushi domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/1179

- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=6901
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/5648
- UniProt
  http://www.uniprot.org/uniprot/P48740

Sources for This Summary

- Atik T, Koparir A, Bademci G, Foster J 2nd, Altunoglu U, Mutlu GY, Bowdin S, Elcioglu N, Tayfun GA, Atik SS, Ozen M, Ozkinay F, Alanay Y, Kayserili H, Thiel S, Tekin M. Novel MASP1 mutations are associated with an expanded phenotype in 3MC1 syndrome. Orphanet J Rare Dis. 2015 Sep 30;10:128. doi: 10.1186/s13023-015-0345-3.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/26419238
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4589207/
- Degn SE, Jensen L, Gál P, Dobó J, Holmvad SH, Jensenius JC, Thiel S. Biological variations of MASP-3 and MAp44, two splice products of the MASP1 gene involved in regulation of the complement system. J Immunol Methods. 2010 Sep 30;361(1-2):37-50. doi: 10.1016/j.jim.2010.07.006. Epub 2010 Jul 29.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20673767
- OMIM: MANNAN-BINDING LECTIN SERINE PROTEASE 1
  http://omim.org/entry/600521
- Rooryck C, Diaz-Font A, Osborn DP, Chabchoub E, Hernandez-Hernandez V, Shamseldin H, Kenny J, Waters A, Jenkins D, Kaissi AA, Leal GF, Dallapiccola B, Carnevale F, Bitner-Glindzicz M, Lees M, Hennekam R, Stanier P, Burns AJ, Peeters H, Alkuraya FS, Beales PL. Mutations in lectin complement pathway genes COLEC11 and MASP1 cause 3MC syndrome. Nat Genet. 2011 Mar; 43(3):197-203. doi: 10.1038/ng.757. Epub 2011 Jan 23.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/21258343
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3045628/
- Sirmaci A, Walsh T, Akay H, Spiliopoulos M, Sakalar YB, Hasanefendioglu-Bayrak A, Duman D, Farooq A, King MC, Tekin M. MASP1 mutations in patients with facial, umbilical, coccygeal, and auditory findings of Carnevale, Malpuech, OSA, and Michels syndromes. Am J Hum Genet. 2010 Nov 12;87(5):679-86. doi: 10.1016/j.ajhg.2010.09.018. Epub 2010 Oct 28.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/21035106
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2978960/
- Yongqing T, Wilmann PG, Reeve SB, Coetzer TH, Smith AI, Whisstock JC, Pike RN, Wijeyewickrema LC. The x-ray crystal structure of mannose-binding lectin-associated serine proteinase-3 reveals the structural basis for enzyme inactivity associated with the Carnevale, Mingarelli, Malpuech, and Michels (3MC) syndrome. J Biol Chem. 2013 Aug 2;288(31):22399-407. doi: 10.1074/jbc.M113.483875. Epub 2013 Jun 21. Erratum in: J Biol Chem. 2013 Sep 27;288(39): 28307.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/23792966
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3829330/

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